Patient Information Sheet
Shortened Long Bones

You have recently learned that your baby has short long bones. We expect that you may have questions about what this could mean for your developing baby. Hopefully, this information will address some of your concerns; however, we encourage you to speak to your genetic counselor or healthcare provider for more information. It is important to remember that anyone can have a baby with a birth defect. Birth defects occur in 2-5% of all births and are rarely caused by something that the parents did or did not do before or during pregnancy. Currently, there are no tests available to detect all health problems.

What is a "short long bone"?

The long bones are the bones that make up our arms and legs. The long bones that are most commonly measured by ultrasound are the humerus in the upper arm and the femur in the upper leg. Based on how far along in the pregnancy you are, there are average long bone measurements that we expect to see when an ultrasound is performed. Your baby's long bone measurements are compared to average long bone measurements of other babies at the same stage in pregnancy. An ultrasound finding of "short long bones" means that one or more of the baby's long bones is measuring shorter than expected. Sometimes phrases like "lagging biometry" or "lagging limb length" are also used to describe shortened long bones.

How many babies are found with "short long bones"?

A short long bone on ultrasound is a common finding seen in up to 6-7% of babies.

What problems could be associated with "short long bones" for my baby?

Most babies with an ultrasound finding of short long bones will be healthy. However, some researchers have found that a baby who has short long bones may have a higher chance for having a chromosome abnormality, such as Down syndrome, a genetic condition, or other pregnancy complications, such as poor fetal growth, later in pregnancy.

In order to understand a possible chromosomal cause or genetic syndrome cause, it is helpful to know a little about chromosomes. Chromosomes are packages of genetic information that carry the instructions (genes) necessary for our growth and development. Typically, there are 23 pairs of chromosomes in each cell of our bodies. One copy of each pair is from our mother and one copy of each pair is from our father. It is important to have the correct amount of genetic material to have typical development. Having an extra or missing whole chromosome, or piece of a chromosome, may cause birth defects. Changes in one or more genes can also result in a particular set of characteristics (also called a syndrome). Chromosome abnormalities and genetic syndromes can produce birth defects and physical changes such as short long bones.

The most common chromosome abnormality seen in babies with shortened long bones is Down syndrome. Down syndrome is caused by having an extra copy of chromosome #21. Individuals with Down syndrome often have characteristics in common, including: a flattened facial appearance, up-slanting eyes, low muscle tone, and short stature. Individuals with Down syndrome typically have mild to moderate intellectual disability and are at an increased risk for medical problems, such as heart and bowel defects. Despite the challenges they may face, children with Down syndrome are more like other children than different. Just like with all children, intellectual and physical abilities in children with Down syndrome cannot be predicted prenatally or even in the early years of life.

The specific chance for your baby to have a chromosome abnormality, other genetic syndrome, or other pregnancy complications will depend on many factors. Your doctor or genetic counselor will discuss these risks with you. Keep in mind, the finding of short long bones *does not* mean that the baby will have noticeably short limbs when he or she is born. In fact, your baby's limbs will most likely look perfectly fine.

What further testing may be offered and what will it tell me?

Depending on your current gestational age, a number of additional tests may be offered to you. If short long bones are found during a routine ultrasound, a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy. Approximately 25-50% of children who have Down syndrome will <u>not</u> have any additional findings on a detailed ultrasound. The ultrasound will only modify your chance of having a baby with a chromosome problem such as Down syndrome. It <u>cannot</u> diagnose or rule out the presence of a chromosomal abnormality.

There are two additional types of prenatal tests: screening tests and diagnostic tests. Screening tests can modify your risk for a chromosome abnormality such as Down syndrome and involve no risk of complications to your pregnancy. A screening test will not determine if your baby has Down syndrome, but it may help you decide whether or not you would like to pursue a diagnostic test.

Diagnostic tests can evaluate the number and structure of your baby's chromosomes. They are 99% accurate in detecting extra or missing chromosomes. Unfortunately, all diagnostic tests have a risk of complications including the potential for pregnancy loss.

Screening Tests

There are multiple screening tests to determine the chances for a pregnancy to have a chromosome condition. These screening tests cannot diagnose or rule out the presence of a chromosome condition but may be used to help you decide whether or not you would like to pursue a diagnostic test.

Non-invasive prenatal testing, or NIPT, is a screening test for certain chromosome conditions. During pregnancy, some of your baby's chromosome material is in your blood, along with your own chromosomes. NIPT is a blood test that measures the amount of chromosome material in your blood to determine if your baby could have an extra chromosome, such as in Down syndrome where there is an extra 21st chromosome. This test also screens for two other more severe chromosome conditions, trisomy 13 and trisomy 18, and may screen for some less severe conditions that are caused by different numbers of the X or Y chromosome. NIPT is a highly accurate screen; however, it is not a diagnostic test. The detection rate for these chromosome conditions is typically between 90-99%.

Other screening blood tests (often called a triple, quad, penta, or first trimester screen) may also be offered. Your genetic counselor or health care provider can discuss these various screening options in more detail.

Diagnostic Tests

Depending on your gestational age, an amniocentesis procedure may be offered. The amniocentesis is a diagnostic test in which a small sample of amniotic fluid is obtained to examine the baby's chromosomes. Because the procedure is considered an invasive procedure, there is a risk, likely less than 1%, for complications that can lead to miscarriage.

If you choose to pursue amniocentesis, you may be offered an additional test called a chromosome microarray (CMA.) It is used to identify small missing or extra pieces of the baby's chromosomes that may be associated with genetic conditions. CMA cannot detect all genetic conditions and may detect variations in the chromosomes that have uncertain clinical significance. Parental blood samples may help to clarify the meaning of a variation, but effects of these changes may not be known until after the baby is born. CMA may also detect information such as non-paternity and close relationships between parents.

The risks, benefits and limitations of screening and diagnostic testing options should be discussed with you by your genetic counselor or other health care provider. Follow-up counseling and referrals for support can be made if a chromosome condition is detected prenatally. As with all situations in which prenatal testing is discussed, it is your decision whether or not this test is done.

What should I do now?

This information is only intended as an introduction to some of the terms and tests that you have already heard or will be hearing about from your genetic counselor or other healthcare provider. We hope that this information sheet will be helpful as you begin to understand more about short long bones. We understand that any time something of concern is found through prenatal screening and testing, parents are going to be worried. Please don't hesitate to contact your genetic counselor with any questions or concerns you have. We are here to help you and your baby.

The decision to have one or more of these tests or to do no additional testing is a difficult and personal one. There is no one right decision. Some people will decide to have no further testing because they do not feel they need to know if their baby will have any of these conditions before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about short long bones, you should make the decision that is right for you and your family. Please keep in mind that most babies with short long bones will not have the complications described in this information sheet. Though it is never possible to predict the long-term prognosis for a child with even isolated short long bones, most babies will be born without significant medical complications.